

What is claimed is:

- 1 1. A substantially purified human small conductance calcium-activated potassium channel
2 -3 (hKCa3/KCNN3) polypeptide comprising an amino acid sequence as set forth in
3 SEQ ID NO:2.
- 1 2. An isolated polynucleotide encoding an hKCa3/KCNN3 polypeptide comprising an
2 amino acid sequence as set forth in SEQ ID NO:2.
- 1 3. An isolated polynucleotide selected from the group consisting of:
2 (a) SEQ ID NO:1, where T can also be a U;
3 (b) nucleic acid sequences complementary to SEQ ID NO:1;
4 (c) fragments of SEQ ID NO:1 that are at least 15 bases in length and will
5 hybridize to DNA which encodes a polypeptide as set forth in SEQ ID
6 NO:2.
- 1 4. The polynucleotide of claim 2, wherein said polynucleotide is operatively linked to an
2 expression control sequence.
- 1 5. The polynucleotide of claim 4, wherein the expression control sequence is a promoter.
- 1 6. The polynucleotide of claim 5, wherein the promoter is tissue specific.
- 1 7. An expression vector containing the polynucleotide of claim 2.
- 1 8. The vector of claim 7, wherein the vector is a plasmid.
- 1 9. The vector of claim 7, wherein the vector is a viral vector.
- 1 10. The vector of claim 9, wherein the viral vector is a retroviral vector.

- 1 11. A host cell containing the vector of claim 7.
- 1 12. The host cell of claim 11, wherein the cell is a eukaryotic cell.
- 1 13. The host cell of claim 11, wherein the cell is a prokaryotic cell.
- 1 14. An antibody which binds to an hKCa3/KCNN3 polypeptide having an amino acid
2 sequence as set forth in SEQ ID NO:2 or conservative variants thereof.
- 1 15. The antibody of claim 14, wherein the antibody is monoclonal.
- 1 16. The antibody of claim 14, wherein the antibody is polyclonal.
- 1 17. A method for identifying a compound which affects hKCa3/KCNN3, comprising:
2 (a) incubating the compound and a sample of interest, wherein said sample
3 contains a member of the group consisting of hKCa3/KCNN3
4 polypeptide and hKCa3/KCNN3 polynucleotide, under conditions
5 sufficient to allow the compound of interest to interact with the sample;
6 (b) determining the effect of the compound on the expression or activity of
7 hKCa3/KCNN3.
- 1 18. The method of claim 17, wherein the sample of interest is a host cell containing an
2 expression vector comprising an isolated polynucleotide encoding the hKCa3/KCNN3
3 polypeptide encoding SEQ ID NO:2.

- 1 19. The method of claim 17, wherein the sample of interest is a cell line expressing an
2 hKCa3/KCNN3 polypeptide.
- 1 20. The method of claim 17, wherein the sample of interest is hKCa3/KCNN3 polypeptide
2 having an amino acid sequence as set forth in SEQ ID NO:2.
- 1 21. The method of claim 17, wherein the sample of interest is a polynucleotide encoding
2 SEQ ID NO:2.
- 1 22. The method of claim 17, wherein the compound is selected from the group consisting
2 of a peptide, peptidomimetic, chemical compound, and a pharmaceutical compound.
- 1 23. A method for diagnosis of a subject having or at risk of having a hKCa3/KCNN3-
2 associated disorder, comprising the steps of :
- 3 (a) contacting a sample from the subject suspected of having or at risk of
4 having a hKCa3/KCNN3-associated disorder with a reagent that binds to
5 hKCa3/KCNN3,
6 (b) detecting binding of the reagent to hKCa3/KCNN3,
7 (c) comparing the binding of the reagent to said sample with the binding of
8 the reagent to a control sample.
- 1 24. The method of claim 21, wherein the sample is nucleic acid.
- 1 25. The method of claim 24, further comprising amplifying the nucleic acid of the sample
2 prior to contacting a sample from the subject suspected of having a hKCa3/KCNN3
3 disorder with a reagent that binds to hKCa3/KCNN3.
- 1 26. The method of claim 21, wherein the sample is a biopsy, blood, plasma, serum, or
2 urine.

1 27. The method of claim 21, wherein the disorder is selected from the group consisting of
2 a neuropsychiatric, neurological, neuromuscular, and immunological disorder.

1 28. The method of claim 21, wherein the disorder is bipolar disease.

1 29. The method of claim 21, wherein the disorder is schizophrenia.

1 30. The method of claim 21, wherein the reagent is an antibody which binds to
2 hKCa3/KCNN3 polypeptide.

1 31. The method of claim 21, wherein the reagent is a polynucleotide which encodes SEQ
2 ID NO:2.

1 32. The method of claim 21, wherein the reagent is detectably labeled.

1 33. The method of claim 32, wherein the detectable label is selected from the group
2 consisting of a radioisotope, a fluorescent compound, a bioluminescent compound and
3 a chemiluminescent compound.

1 34. A method of diagnosis of a subject having a hKCa3/KCNN3-associated or at risk of
2 having a hKCa3/KCNN3-associated disorder comprising:

3 (a) identifying the presence of a trinucleotide repeat in the 5'-coding region
4 of the hKCa3/KCNN3 gene; and

5 (b) comparing the trinucleotide repeat region from the subject with the same
6 region from a normal subject or a standard sample, thereby providing a
7 diagnosis of the subject.

1 35. The method of claim 34, wherein the disorder is selected from the group consisting of
2 a neuropsychiatric, neurological, neuromuscular, and immunological disorder.

- 1 36. The method of claim 34, wherein the disorder is schizophrenia.
- 1 37. The method of claim 34, wherein the disorder is bipolar disease.
- 1 38. A method of determining the prognosis of a subject with an hKCa3/KCNN3-associated
2 disorder, comprising:
3 (a) obtaining a sample from said subject,
4 (b) determining the number of a trinucleotide CAG/CTG repeats in the
5 5'-coding region of the hKCa3/KCNN3 gene;
6 (c) correlating the number of a trinucleotide repeats in the 5'-coding region
7 of the hKCa3/KCNN3 gene in said subject with the prognosis of the
8 subject.
- 1 39. The method of claim 38, wherein the disorder is selected from the group consisting of
2 a neuropsychiatric, neurological, neuromuscular, and immunological disorder.
- 1 40. The method of claim 38, wherein said disorder is schizophrenia.
- 1 41. The method of claim 38, wherein said disorder is bipolar disease.
- 1 42. The method of claim 38, wherein said sample consists of a biopsy, blood, plasma, or
2 urine sample obtained from said subject.

- 1 43. The method of determining a treatment regimen of neuropsychiatric, neurological,
2 neuromuscular or immunological disorders in a subject with an hKCa3/KCNN3-
3 associated disorder, comprising:
- 4 (a) obtaining a sample from said subject,
 - 5 (b) determining the number of a trinucleotide CAG/CTG repeats in the
6 5'-coding region of the hKCa3/KCNN3 gene;
 - 7 (c) correlating the number of a trinucleotide repeats in the 5'-coding region
8 of the hKCa3/KCNN3 gene in said subject with the prognosis of the
9 subject.
- 1 44. The method of claim 43, wherein the disorder is selected from the group consisting of
2 a neuropsychiatric, neurological, neuromuscular, and immunological disorder.
- 1 45. The method of claim 43, wherein said disorder is schizophrenia.
- 1 46. The method of claim 43, wherein said disorder is bipolar disease.
- 1 47. The method of claim 43, wherein said sample consists of a biopsy, blood, plasma, or
2 urine sample obtained from said subject.
- 1 48. A method for determining the prognosis of a subject diagnosed with an
2 hKCa3/KCNN3-associated disorder, comprising:
- 3 obtaining a sample from said subject,
 - 4 determining the alleles of hKCa3 expressed in said sample from said subject,
 - 5 and
 - 6 correlating the alleles with the prognosis of said subject.
- 1 49. The method of claim 48, wherein the disorder is selected from the group consisting of
2 a neuropsychiatric, neurological, neuromuscular, and immunological disorder.

- 1 50. The method of claim 48, wherein said disorder is schizophrenia.
- 1 51. The method of claim 48, wherein said disorder is bipolar disease.
- 1 52. The method of claim 48, wherein said sample is a member selected from the group
2 consisting of a biopsy, blood, plasma, or urine sample
- 1 53. A method for treating a subject having or at risk of having an hKCa3/KCNN3-
2 associated or hKCa3/KCNN3-related disorder, comprising administering to the subject
3 a therapeutically effective amount of a polypeptide of SEQ ID NO:2.
- 1 54. The method of claim 53, wherein the disorder is selected from the group consisting of
2 a neuropsychiatric, neurological, neuromuscular, and immunological disorder.
- 1 55. A method of treating a subject having or at risk of having an hKCa3/KCNN3-
2 associated or hKCa3/KCNN3-related disorder, comprising administering to the subject
3 a therapeutically effective amount of a polynucleotide encoding SEQ ID NO:2.
- 1 56. The method of claim 55, wherein the disorder is selected from the group consisting of
2 a neuropsychiatric, neurological, neuromuscular, and immunological disorder.
- 1 57. A method of treating a patient having or at risk of having an hKCa3/KCNN3-
2 associated or hKCa3/KCNN3-related disorder, the method comprising:
3 introducing into a cell of a patient having an hKCa3/KCNN3-associated
4 disorder a nucleotide sequence encoding a SEQ ID NO:2 and a eukaryotic
5 promoting sequence operably linked thereto, said introducing resulting in the
6 genetic transformation of the cell so that the nucleotide sequence expresses
7 SEQ ID NO:2.

- 1 58. The method of claim 57, wherein the disorder is selected from the group consisting of
2 a neuropsychiatric, neurological, neuromuscular, and immunological disorder.
- 1 59. A composition for administration of hKCa3/KCNN3 to a patient having an
2 hKCa3/KCNN3-associated or hKCa3/KCNN3-related disorder comprising:
3 (a) a therapeutically effective amount of a substantially pure
4 hKCa3/KCNN3 polypeptide; and
5 (b) a pharmaceutically acceptable carrier.
- 1 60. The composition of claim 55, wherein the carrier is a liposome.
- 1 61. A kit useful for detecting the presence of hKCa3/KCNN3 in a sample from a subject
2 having a hKCa3/KCNN3-associated or hKCa3/KCNN3-related disorder, the kit
3 comprising: carrier means being compartmentalized to receive in close confinement
4 therein one or more containers comprising a container containing an antibody which
5 specifically binds to hKCa3/KCNN3.
- 1 62. A kit useful for the detection of a target nucleic acid sequence in a sample from a
2 subject having a hKCa3/KCNN3-associated or hKCa3/KCNN3-related disorder,
3 wherein the presence of the target nucleic acid sequence in the sample is indicative of
4 having or predisposed to having a human hKCa3/KCNN3-associated disorder, the kit
5 comprising: carrier means being compartmentalized to receive in close confinement
6 therein one or more containers comprising a container containing oligonucleotides
7 which hybridize to hKCa3/KCNN3 nucleic acid sequences.
- 1 63. A transgenic nonhuman animal having a phenotype characterized by expression of
2 hKCa3/KCNN3, otherwise not naturally occurring in the animal, the phenotype being
3 conferred by a transgene contained in the somatic and germ cells of the animal, the
4 transgene comprising a nucleic acid sequence which encodes hKCa3/KCNN3.

- 1 64. The transgenic nonhuman animal of claim 63, wherein the animal is a mouse.